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Letter to the Editor

Geleophysic Dysplasia vs. Myhre Syndrome

To the Editor:

The paper by Rosser et al. [1995] reports on geleophysic dysplasia (GD), a rare autosomal-recessive entity (MIM *231050) [McKusick, 1994], first described by Spranger et al. The first two patients presented by Rosser et al. [1995] had typical manifestations of GD, particularly regarding facial characteristics. The fact that they were brothers, from unaffected parents, supported autosomal-recessive inheritance. However, a careful analysis of the third patient demonstrated differences from patients 1 and 2, and I suggest Myhre syndrome (MS) rather than GD be considered as diagnosis.

MS is another rare growth-mental deficiency syndrome, and only 5 cases have been reported so far; autosomal-dominant inheritance has been proposed (MIM 139210) but not confirmed, and de novo mutations have been invoked in all cases [Myhre et al., 1981; Soljak et al., 1983; García-Cruz et al., 1993]. Many findings of the above patient seem to correspond to MS, including: short palpebral fissures, hearing loss, short stature, low weight, OFC <3rd centile, and decreased joint mobility. On the other hand, the case is sporadic without parental consanguinity, as is true of the previously reported MS patients. Additionally, he did not present the characteristic depressed nasal bridge and/or cardiac involvement of GD patients, and finally, the patient's facial picture gives a "gestalt" that corresponds to MS.

It is clear that the GD and MS share many manifestations; nevertheless, the distinction between them seems not too difficult. Probably because of its rarity, MS was not considered by Rosser et al. [1995]. So far, no molecular defect has been found in either GD or MS, but it would not be surprising if they turned out to be caused by mutations of the same gene, as has been demonstrated in the craniosynostoses caused by mutations in fibroblast growth factor receptor 2 (FGFR2) [Cohen, 1995; Muenke and Schell, 1995].

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